AN UNUSUAL CASE OF CONGENITAL HEART ANOMALY IN A WOMAN AGED TWENTY-FIVE YEARS

LINDEN F. EDWARDS, PH.D. AND IRMA EGLITIS, M.D.
Department of Anatomy, The Ohio State University, College of Medicine, Columbus 10

During the routine dissection in the Human Anatomy Laboratory, Ohio State University, College of Medicine, our attention was called by the students at one of the tables to the presence of an abnormal heart and arrangement of the great vessels in the cadaver they were dissecting. At our request the heart and remnants of the great vessels were preserved for future detailed study. Unfortunately during the removal of the specimen many of the vessels were not as well preserved as they should have been. It is on this material that we present the following description.

A unique feature of the findings is the age of the individual. According to the death certificate this person was a female born June 6, 1926 and died October 17, 1951, at the age of twenty-five. According to the clinical report obtained from the Orient State Institute, where she had been a patient since July 3, 1941, "her death was sudden. She was found to have a congenital heart anomaly, with clubbing of the fingers and toes, dyspnea and cyanosis. She had a mental age of two years with an I.Q. of 14. She was about 5′4″ in height and weighed 120 pounds."

ANATOMICAL DESCRIPTION OF HEART

The external appearance of the heart can be seen in figure A, and it is to be noted that it is conical in shape without a well-marked apex. Its weight and measurements are normal.

The typical four chambers are present, although greatly modified. Thus, for example the auricle of the right atrium is enlarged and is more quadrilateral-shaped than the typical ear-shape. The openings of the venae cavae are normal. The (Eustachian) valve of the inferior vena cava (fig. C) is also normal in size and appearance, whereas, the Thebesian valve is quite pronounced in conformity with the markedly enlarged opening of the coronary sinus. A fossa ovalis is barely visible and there is no sign of a foramen ovale.

The cavity of the right ventricle (figs. C and D) is quite noticeably smaller than normal and its walls are greatly hypertrophied. The tricuspid valve is

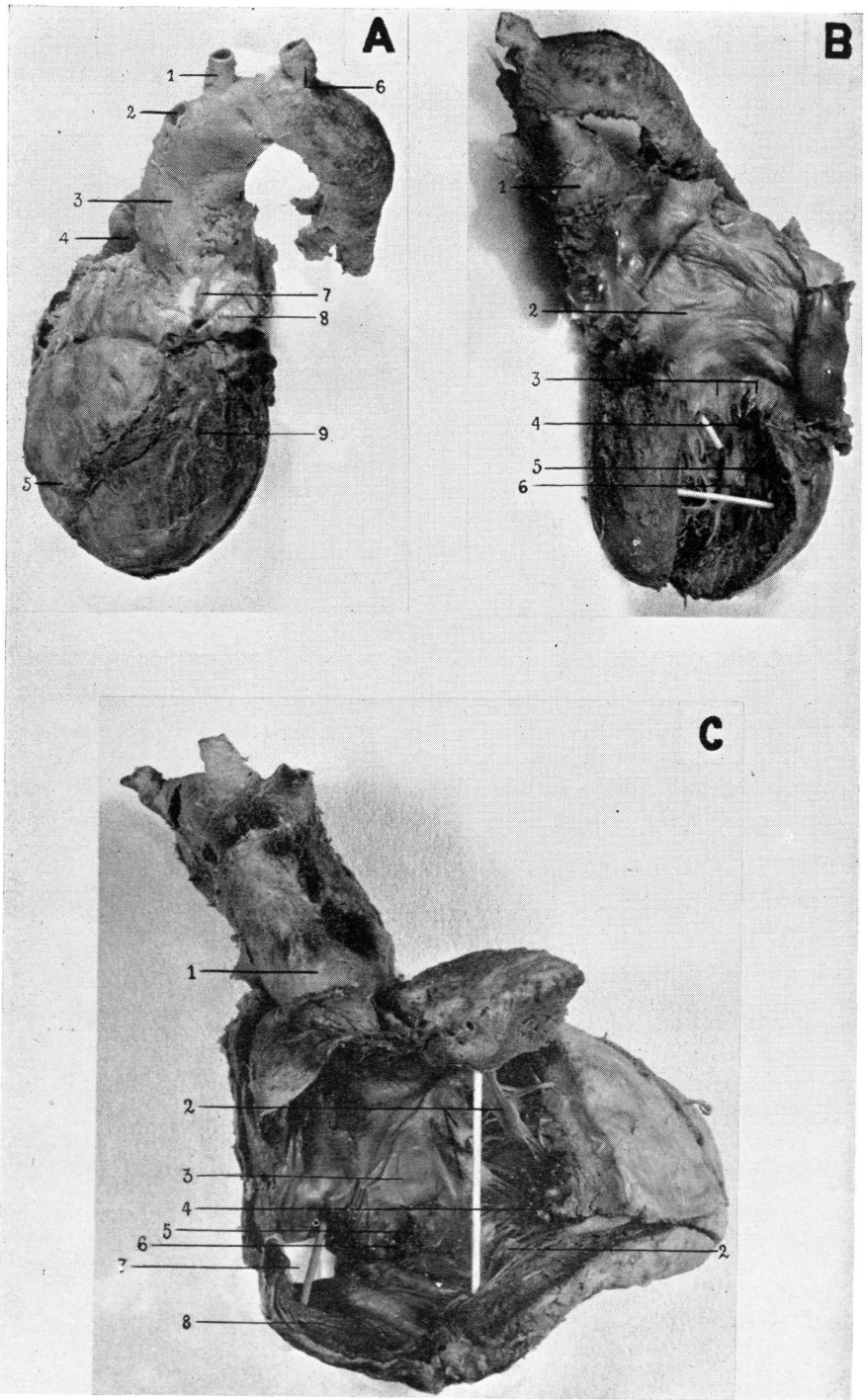
---

EXPLANATION OF FIGURES IN PLATE I

FIGURE A. In this figure the heart is shown more or less in the normal position, for ease of orientation and, therefore, shows the aortic arch to the left. Whereas, in the cadaver, the ventricles pointed to the left, the atria to the right and the aortic arch was also directed to the right (See Figs. C and E). 1—Right com. carotid art.; 2—Innominate art.; 3—Truncus arteriosus; 4—Right auricle; 5—Right ventricle; 6—Right subclavian art.; 7—Ligamentous structure; 8—Left auricle; 9—Left ventricle.

FIGURE B. In this figure the heart is also shown in a vertical position but is rotated to the right in order to expose the left chambers. 1—Truncus arteriosus; 2—Cavity of the left atrium; 3—Bicuspid valve; 4—Chordae tendineae; 5—Cavity of the left ventricle; 6—Papillary muscle.

FIGURE C. The heart in this figure is oriented according to the position it assumed in the cadaver, i.e. with the ventricles pointing toward the left, the atria toward the right and the aortic arch directed toward the right. 1—Truncus arteriosus; 2—Tricuspid valve cusps; 3—Cavity of the right atrium; 4—Cavity of the right ventricle; 5—Atrial opening of the coronary sinus; 6—Thebiesian valve of coronary sinus; 7—Opening of the sup. vena cava; 8—Eustachian valve of inf. vena cava.


A
1
2
3
4
5
6
7
8
9
B
1
2
3
4
5
6
C
1
2
3
4
5
6
7
8
2

normal. It is to be noted (fig. A) that a conus arteriosus and pulmonary trunk are absent.

The auricle of the left atrium is of typical form (fig. A), although it is somewhat more triangular in shape than normal. The left atrium is abnormally large and extends to the right, posterior to the right atrium (fig. B), its right margin being located at the level of the atrial orifice of the superior vena cava. A single right pulmonary vein and two left pulmonary veins empty into the left atrium.

The left ventricle (figs. A and B) is normal but is much larger than the right. Its volume in comparison with that of the right ventricle is more than doubled. The mitral valve is normal.

The interventricular septum is deficient at its cranial end, due to the failure of its membranous portion to develop, as a result of which a communication exists between the two ventricles (fig. D). Correlated with this deficiency and with the absence of a pulmonary trunk, is the presence of a persistent single arterial trunk, the truncus arteriosus (figs. A-D). Obviously this single trunk serves to conduct the venous blood from the right side of the heart and the arterial blood from the left side.

TRUNCUS ARTERIOSUS

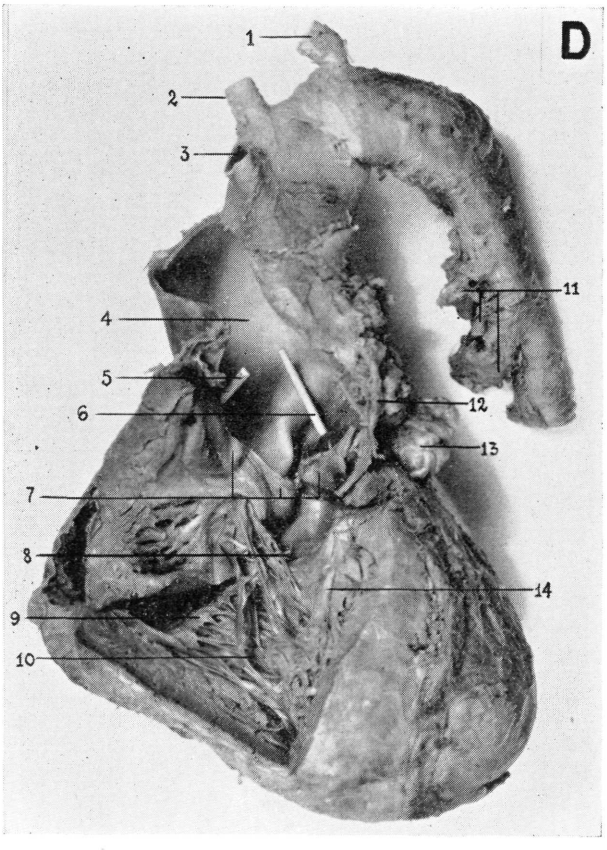
The orifice of the truncus arteriosus (fig. D) over-rides the septal defect and is situated slightly in front and to the left of the septal cusp of the tricuspid valve and directly to the right of the anterior cusp of the mitral valve. Thus, as regards its relation to the latter cusp its situation is similar to that of the aortic vestibule of the normal heart. The opening is provided with three well-developed semilunar cusps, each having a pronounced nodule (corpus Arantii). The cusps are located one on the right, one on the left and one posteriorly, the margin of the latter being roughened. The sinuses (of Valsalva) are large. In the right sinus is the opening of the right coronary artery, which seems to be unusually wide, and cranial to the left sinus is the opening of the left coronary artery. The latter opening is directed obliquely and is provided with a slight valve-like fold. Dissection of the coronary arteries proves them to take a normal course and to have typical branches of distribution.

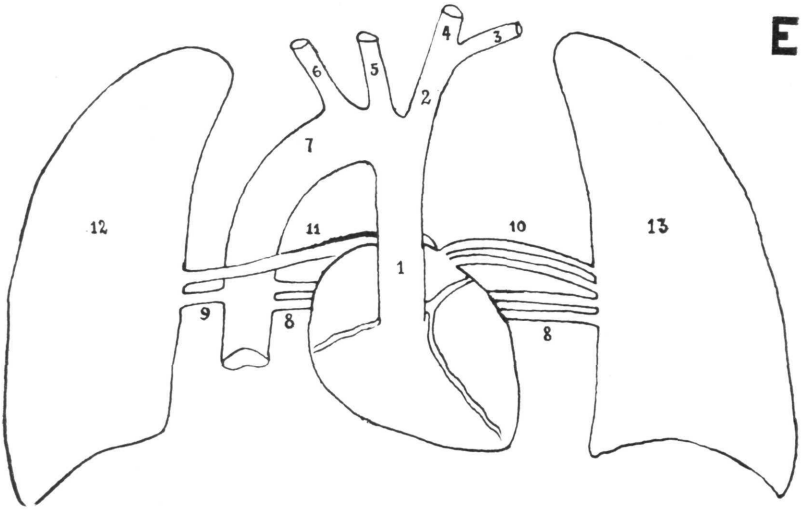
Unfortunately no radiographs of this patient's chest were available and it is, therefore, difficult to determine the exact position which the heart and truncus arteriosus assumed in the living state. However, figure E, which was furnished to the authors by the students who dissected the body, shows that the arch of the truncus curved to the right rather than to the left and that the descending portion was situated on the right side of the thoracic cavity. It can also be seen in figure E that the branches of the arch are the reverse of the normal. Figures A, B, D fail to show these features because the heart and truncus arteriosus were photographed in a vertical position for ease of orientation.

EXPLANATION OF FIGURES IN PLATE II

FIGURE D. In this figure the heart and aortic arch are oriented in the normal position. However, in the cadaver it assumed the position as that shown in figures C and E. 1—Right subclavian art.; 2—Right com. carotid art.; 3—Innominate art.; 4—Truncus arteriosus (laid open); 5—Probe in right coronary art.; 6—Probe in left coronary art.; 7—Semilunar cusps; 8—Communication between the right and left ventricle; 9—Tricuspid valve; 10—Cavity of right ventricle; 11—Bronchial arteries; 12—Ligamentous structure; 13—Left auricle; 14—Interventricular septum.

FIGURE E. Schematic representation of the heart and vessels as actually found in the cadaver. 1—Truncus arteriosus; 2—Innominate art.; 3—Left subclavian art.; 4—Left common carotid art.; 5—Right common carotid art.; 6—Right subclavian art.; 7—Aortic arch; 8—Left bronchial arteries; 9—Right bronchial artery; 10—Left pulmonary veins; 11—Right pulmonary vein; 12—Right lung; 13—Left lung.


D
1
2
3
4
5
6
7
8
9
10
11
12
13
14


E
1
2
3
4
5
6
7
8
8
9
10
11
12
13

Situated on the ventral aspect of the proximal end of the truncus arteriosus (figs. A and D) is a structure that is ligamentous proximally and bifurcates distally into two branches, each of which has a lumen. Unfortunately this structure is not illustrated in figure E and it is, therefore, impossible to determine what the original distribution of the two branches was before they were severed at the time the heart was removed from the body. The proximal end fuses with the wall of the truncus arteriosus inferior to the left semilunar cusp and the question arises as to whether this ligamentous structure should be considered an atretic pulmonary trunk.

Compensating for the absence of pulmonary arteries are three greatly dilated bronchial arteries, which branch from the descending portion of the truncus arteriosus, one to the right lung and two to the left lung (figs. D and E). They correspond in number with the pulmonary veins.

There is no evidence of a ligamentous ductus arteriosus. However, a well-defined vestigial fold of Marshall is present. In the absence of a left pulmonary artery it extends from the region of the left bronchial arteries to the left pulmonary veins, where the latter empty into the left atrium. Cardiac veins present a normal pattern and drain into the coronary sinus which is markedly enlarged (fig. C).

DISCUSSION

The question arises as to the classification of the present cardiac anomaly. Obviously it cannot be diagnosed as Tetralogy of Fallot, inasmuch as the latter is characterized, among other cardiac defects, by pulmonary stenosis. Pulmonary arteries are absent in the present case. According to some authorities, notably Abbott (1936), Humphrey (1932) and Kugel (1931), the condition known as Truncus Arteriosus Communis Persistens is characterized by the presence of a single large arterial trunk leading from the heart and giving rise to coronary, pulmonary and systemic vessels, by a high interventricular defect, by a patent foramen ovale and by a tremendously enlarged heart, especially the right ventricle. Moreover, as Taussig (1947) points out, in cases of a persistent pulmonary artery arising from a common truncus arteriosus there is adequate circulation to the lungs, a large volume of oxygenated blood is mixed with venous blood and, therefore, cyanosis is minimal or absent.

Opposed to the "true" form of truncus arteriosus is the so-called "pseudo" form, or as it is sometimes called, Truncus Solitarius Aorticus. The latter condition is characterized by atresia of the pulmonary artery, due to the failure of the sixth aortic arches to develop, only a vestige of which persists as a ligamentous structure ending blindly at the heart. In this condition circulation to the lungs can only be by way of bronchial arteries, which become greatly enlarged, in order to accommodate the large volume of blood. According to Taussig (1947) the bronchial arteries are never sufficiently great to permit adequate circulation to the lungs; a small volume of oxygenated blood is mixed with a large volume of venous blood and consequently cyanosis is intense.

Clubbing of the distal phalanges of the fingers and toes in congenital heart disease is claimed to be proportional to the degree of cyanosis present and is related to a state of chronic passive congestion of the capillaries of the nail bed. Cyanosis is always manifested before clubbing, the earliest sign of the latter being a thickening of the nail beds of the index fingers and thumbs. The skin covering the distal phalanges of the digits becomes smooth and shiny and of a lilac hue, while at the same time the base of the nail gradually becomes raised, the basal angle of the nail and digit is obliterated, increased curvature of the nail in both directions gradually develops and finally the tips of the digits become broadened resembling a drumstick in appearance. This condition is attributed indirectly to the low degree of oxygenation of the blood which is due in part to the admixture

of arterial and venous blood from both ventricles prior to its passage into the single arterial trunk and in part to the amount of blood delivered to the lungs for aeration. When circulation to the lungs is by way of bronchial arteries, even though they are enlarged, as in the present described case, the pulmonary blood flow is inadequate, and, therefore, there is intense cyanosis, with peripheral stasis in the capillaries which become dilated. Clubbing of the fingers and toes is attributed directly to passive congestion of the capillaries of the nail-bed, to edema of the connective tissue between the nail-bed and the bone and to the resultant anoxemia, sluggish circulation, increased local deoxygenation and accumulation of metabolites (Brown, 1950).

In view of the anatomical findings and case history of the patient it would seem, therefore, that the present cardiac anomaly may be diagnosed as Truncus Solitarius Aorticus. This type of cardiac defect is not uncommon. However, the present case is unique because of the survival of the patient to the age of twenty-five. Taussig (1947) claims that cardiac anomalies of this sort are incompatible with life, usually the prognosis is extremely poor and "the vast majority of infants die within the first few weeks of life." Schnitker (1952) likewise points out that the prognosis is generally grave but is somewhat better in those cases in which pulmonary arteries arise directly from the common arterial trunk—"In such cases individuals have survived to the age of 18, 22, 25 and 33 years." One of Abbott's cases (1927) in the latter category lived to be 25 and Carr, Goodale and Rockwell (1935) described a patient who survived to the age of 36 years and two months.

A thorough review of the literature revealed but one case closely similar to the present one that survived for as long as 25 years. That case was reported by Zimmermann (1927), who incidentally classified it as truncus arteriosus communis, notwithstanding his claim that pulmonary arteries were absent. His case differed from the present one in the following particulars: there was no clubbing of the terminal phalanges, the common arterial trunk arose from the left side of the base of the heart, the cavity of the right atrium was approximately three times the size of the left, the three branches of the aortic arch arose in the normal manner, the descending aorta was on the left side, there were two pulmonary veins which united to form a common trunk from each lung and there were three right and two left bronchial arteries arising from the descending aorta.

SUMMARY AND CONCLUSIONS

1. An unusual anomalous heart found in a twenty-five year old female cadaver in the Human Anatomy Laboratory, Ohio State University, is described.

2. The most outstanding morphological features of this heart are (a) the absence of a conus arteriosus and pulmonary trunk; (b) the presence of a single large arterial trunk which combines the functions of a systemic and pulmonary circulation; (c) greatly dilated bronchial arteries arising from the descending aorta is the only means whereby blood reaches the lungs; (d) there is a high interventricular septal defect, at which point the orifice of the single arterial trunk, guarded by three semilunar valves, is located; (e) there is no evidence of a ligamentum ductus arteriosus or fossa ovalis; (f) a ligament situated on the ventral aspect and parallel with the single arterial trunk suggests an atretic pulmonary artery; (g) the aortic arch and descending aorta are on the right rather than on the left side, the three branches of the arch being the reverse of normal; (h) the right auricle and the opening of the coronary sinus in the right atrium are enlarged, the latter space being normal in size; (i) the right ventricle has less than half the capacity of the left ventricle, its walls being greatly hypertrophied; (j)the cavity of the left atrium is more spacious than that of the right atrium, and (k) its weight and measurements are normal.

3. In view of the clinical history of the patient and of the anatomical findings in the present described case of cardiac anomaly it is concluded that the anomaly

should be classified as Truncus Solitarius Aorticus. Although this condition is not uncommon it is incompatible with life and usually terminates fatally within a few weeks following birth. The rarity of the described case is confirmed by a review of the literature which reveals only one other reported case of the same survival period, the anatomical findings, however, were not comparable in all details.

BIBLIOGRAPHY

Abbott, M. E. 1936. Atlas of congenital cardiac disease. Persistent truncus arteriosus. Amer. Heart Association, New York. p. 52.

Beaver, D. C. 1933. Persistent truncus arteriosus and congenital absence of one kidney with other developmental defects. Arch. Path., 15: 51–54.

Black, D. D. 1914. Two cases of cardiac malformation—more especially of the infundibular region. Jour. Anat. and Physiol., 48: 274–279.

Brown, J. W. 1950. Congenital heart disease. Staples Press, New York.

Carr, F. B., R. H. Goodale, and **A. E. P. Rockwell.** 1935. Persistent truncus arteriosus in a man aged thirty-six years. Arch. Path., 19: 833–837.

Dickson, W. E. C., and **J. Fraser.** 1914. A congenital abnormality of the heart and blood-vessels. Jour. Anat. and Physiol., 48: 210–214.

Dry, T. J., (and 5 others). 1948. Congenital anomalies of the heart and great vessels. Charles C. Thomas Publisher, Springfield, Ill. Pp. 46–47.

Edwards, J. E. 1953. Congenital malformations of the heart and great vessels. In Pathology of the Heart. S. E. Gould. Charles C. Thomas Publisher, Springfield, Ill. Pp. 266–368.

———., (and 5 others). 1954. An Atlas of Congenital Anomalies of the Heart and Great Vessels. Charles C. Thomas Publisher, Springfield, Ill.

Feller, A. 1931. Zur Kenntnis der angeborenen Herzkrankheiten. 1. Mitteilung. Truncus arteriosus communis persistens und seine formale Entstehung. Virch. Arch. f. pathol. Anat. u. Phys., 279: 869–910.

Finley, K. H. 1930. A congenital anomaly of the heart. Truncus arteriosus communis with subacute endocarditis. Amer. Jour. Path., 3: 317–323.

Gross, R. E. 1947. Surgical Treatment for Abnormalities of the Heart and Great Vessels. Charles C. Thomas Publisher, Springfield, Ill.

Halpert, B., and **R. Tennant.** 1934. Anomalies of the interventricular septum and pulmonary orifice. Amer. Jour. Path., 3: 375–382.

Hülse, W. 1918. Beiträg zur Kenntnis der totalen Persistenz des Truncus arteriosus communis. Virch. Arch. f. pathol. Anat. u. Phys., 225: 16–23.

Humphreys, E. M. 1932. Truncus arteriosus communis persistens. Arch. Path., 14: 671–700.

Klemke, W. 1926. A classic case of total persistence of the truncus arteriosus. Abst., Arch. Path. and Lab. Medic., 1: 136.

Knape, W. 1912. Eine seltene Herzmissbildung bei Situs inversus abdominis. Virch. Arch. f. path. Anat. u. Phys., 209: 473–476.

Kugel, M. A. 1931. A clinical and pathological study of two cases of truncus solitarius aorticus (Pulmonary Atresia). Amer. Heart Jour., 7: 262–273.

Mason, D. G., and **W. C. Hunter.** 1937. Localized congenital defects of the cardiac interventricular septum. Amer. Jour. Path., 5: 835–843.

Miller, M. K., and **M. W. Lyon, Jr.** 1931. Persistent truncus arteriosus: Cardiac hypertrophy, dysphagia, death on eleventh day. Amer. Heart Jour., 7: 106–109.

Ngai, S. K. 1935. Congenital anomaly of the heart. Amer. Jour. Path., 2: 309–321.

Plesch, J. 1937. Physiology and pathology of the heart and blood-vessels. Oxford University Press, London.

Schnitker, M. 1952. Congenital anomalies of the heart and great vessels. Truncus arteriosus communis persistens. Oxford University Press, New York. Pp. 252–258.

Taussig, H. B. 1947. Congenital malformations of the heart. Truncus arteriosus. The Commonwealth Fund, New York. Pp. 247–277.

Webb, A. C. 1946. Truncus arteriosus communis persistens. Arch. Path., 42: 427–432.

Wenner, O. 1909. Beiträge zur Lehre der Herzmissbildungen. Virch. Arch. f. path. Anat. u. Phys., 196: 127–167.

Zimmermann, H. M. 1927. A Congenital anomaly of the heart: Truncus arteriosus communis. Amer. Jour. Path., 3: 617–622.